



DPY19L2 gene

dpy-19 like 2

Normal Function

The *DPY19L2* gene provides instructions for making a protein that is found in developing sperm cells. The DPY19L2 protein plays a role in the development of the acrosome, a cap-like structure in the head of sperm cells. The acrosome contains enzymes that break down the outer membrane of egg cells, allowing the sperm to fertilize an egg.

The developing acrosome is attached to the nucleus of the cell. The DPY19L2 protein, which is found within the membrane of the nucleus, helps attach the forming acrosome to the nuclear membrane. As the acrosome develops and the sperm cell matures, the acrosome moves to the tip of the head of the sperm, which helps the head elongate into an oval shape.

Health Conditions Related to Genetic Changes

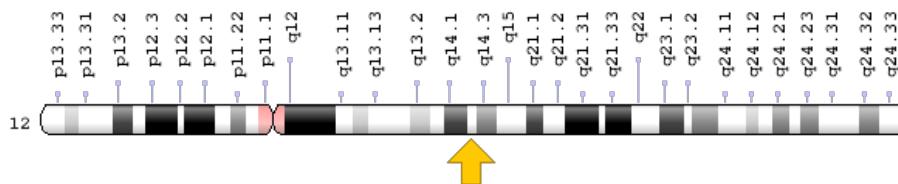
globozoospermia

At least 17 *DPY19L2* gene mutations have been found in men with globozoospermia, a condition characterized by abnormal sperm cells that have a round head and no acrosome. Approximately 70 percent of men with this condition have mutations in *DPY19L2*. Most of these mutations delete large regions of the gene or the whole gene. Others change single protein building blocks (amino acids) in the DPY19L2 protein. These mutations lead to a loss of functional DPY19L2 protein. Without this protein, the forming acrosome is not attached to the nucleus and is removed from the cell. As a result, sperm cells have no acrosome and the head of the sperm does not elongate. The abnormal sperm are unable to get through the outer membrane of an egg cell to fertilize it, leading to an inability to father biological children (infertility) in affected men. Researchers have described other characteristics of the abnormal sperm cells that make fertilization of an egg cell difficult, although it is not clear how changes in the *DPY19L2* gene are involved in development of these characteristics.

Chromosomal Location

Cytogenetic Location: 12q14.2, which is the long (q) arm of chromosome 12 at position 14.2

Molecular Location: base pairs 63,558,913 to 63,669,201 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- D19L2_HUMAN
- dpy-19 like 2 (*C. elegans*)
- dpy-19-like 2 (*C. elegans*)
- FLJ32949
- probable C-mannosyltransferase DPY19L2
- protein dpy-19 homolog 2
- SPATA34
- spermatogenesis associated 34
- SPGF9

Additional Information & Resources

Educational Resources

- Developmental Biology (sixth edition, 2000): Figure 7.2 The Modification of a Germ Cell to Form a Mammalian Sperm
<https://www.ncbi.nlm.nih.gov/books/NBK10005/figure/A1345/?report=objectonly>
- Developmental Biology (sixth edition, 2000): Spermiogenesis
<https://www.ncbi.nlm.nih.gov/books/NBK10095/#A4693>
- Molecular Biology of the Cell (fourth edition, 2002): Sperm
<https://www.ncbi.nlm.nih.gov/books/NBK26914/>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28DPY19L2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- DPY19-LIKE 2
<http://omim.org/entry/613893>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_DPY19L2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=DPY19L2%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=19414
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/283417>
- UniProt
<http://www.uniprot.org/uniprot/Q6NUT2>

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